PACS1 gene

phosphofurin acidic cluster sorting protein 1

Normal Function

The *PACS1* gene provides instructions for making a protein called phosphofurin acidic cluster sorting protein 1 (PACS1). The PACS1 protein is found in a complex network of membranes known as the trans-Golgi network, which sorts proteins and other molecules and sends them to their intended destinations inside or outside the cell. Within the trans-Golgi network, this protein helps transport certain molecules and proteins. The PACS1 protein is most active during development before birth.

Health Conditions Related to Genetic Changes

PACS1 syndrome

At least two mutations in the *PACS1* gene have been found to cause *PACS1* syndrome. This condition is characterized by intellectual disability, speech and language problems, and a distinct facial appearance. Many affected individuals have additional neurological, behavioral, and health problems. The most common mutation, which occurs in nearly everyone with *PACS1* syndrome, results in the production of a protein with the protein building block (amino acid) arginine replaced with the amino acid tryptophan at position 203 (written as Arg203Trp or R203W).

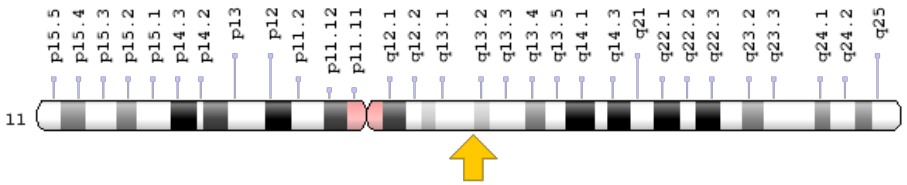
PACS1 gene mutations are thought to impair the protein's ability to aid in the transport of molecules and proteins. Such an impairment likely results in the accumulation or misplacement of these substances within cells. The accumulated molecules and proteins may interfere with the function of the protein produced from the normal copy of the *PACS1* gene, further disrupting the placement of these substances.

Research suggests that impaired PACS1 protein function disrupts normal development of structures in the face, leading to a distinct facial appearance. It is likely that the development of other body systems are similarly affected by impaired PACS1 protein function, leading to other signs and symptoms of *PACS1* syndrome, but more research is needed to understand the mechanisms.

Chromosomal Location

Cytogenetic Location: 11q13.1-q13.2, which is the long (q) arm of chromosome 11 between positions 13.1 and 13.2

Molecular Location: base pairs 66,070,272 to 66,244,744 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cytosolic sorting protein PACS-1, human
- FLJ10209
- KIAA1175
- PACS-1
- phosphofurin acidic cluster sorting protein 1, human

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: TGN and Post-Golgi Trafficking https://www.ncbi.nlm.nih.gov/books/NBK6398/#A74069
- Molecular Biology of the Cell (fourth edition, 2002): Transport from the Trans Golgi Network to the Cell Exterior: Exocytosis https://www.ncbi.nlm.nih.gov/books/NBK26892/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PACS1%5BTIAB%5D%29+OR+%28PACS-1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PHOSPHOFURIN ACIDIC CLUSTER SORTING PROTEIN 1
  http://omim.org/entry/607492

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PACS1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PACS1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:30032
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:55690
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/55690
- UniProt
  https://www.uniprot.org/uniprot/Q6VY07

Sources for This Summary

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